virilising adrenal tumours to dexamethasone was a poor diagnostic discriminating criteria for malignancy, showing either suppression or no change in androgen secretion. Our findings suggest that adrenal tumours may differentiate over time.

Our patient shares a partial phenotype of BWS and has a familial history of neuroblastoma, two conditions known to be associated with an increased risk of cancer and suggestive of a genetic origin. The relationship between late embryonic growth and tissue maturation to childhood neoplasia is not well established. However, BWS is known to be a multigenic disorder with dysregulation of the expression of imprinted genes involved in the growth and cell cycle control in the 11p15 chromosomal region (IGF-II, H19,H-ras-1, $P57^{kip2}$). Loss of somatic heterozygosity for alleles on chromosome 11p15.5 has been also proposed as one of the mechanisms involved in the development of adrenocortical tumours [4].

Close follow-up of patients with unilateral adrenal tumours is essential in particular if atypical cortical cells are present outside the tumour. Molecular analysis for improving diagnosis and elucidating the correlation between the clinical phenotypes and genotypes are important in such cases.

References

1. Carr B, Rainey WE, Mason JJ (1986) The role of calcium in steroidogenesis in foetal zone cells of the human foetal adrenal gland. J Clin Endocrinol Metab 63: 913–917
2. Drut RM, Drut R, Gilbert-Barness E, Sotelo-Avila C (1993) Adrenal hyperplastic nodules in Wiedemann-Beckwith syndrome. Birth Defects Orig Art Ser 29: 367–72
3. Gabrilove JL, Seman AT, Sabet R, Mitty HA, Nicolis GL (1981) Virilizing adrenal adenoma with studies on the steroid content of the adrenal venous effluent and a review of the literature. Endocr Rev 2: 462–470
4. Henry I, Jeanpierre M, Couillin P, Barichard F, Serre JL, Journel H, Lamouroux A, Turleau C, de Grouchy J, Junien C (1989) Molecular definition of the 11p15.5 region involved in Beckwith-Wiedemann syndrome and probably predisposition to adrenocortical carcinoma. Hum Genet 81: 273–277
5. Schneid H, Seurin D, Vasquez MP, Gourmelen M, Cabrol S, Le Bouc Y (1993) Parental allele specific methylation of the human insulin-like growth factor II gene and Beckwith-Wiedemann syndrome. J Med Genet 30: 353–362
6. Sherman FE, Bass LW, Fetterman GH (1958) Congenital metastasizing adrenal cortical carcinoma associated with cytomegaly of the foetal adrenal cortex. Am J Clin Pathol 30: 439–446

V. Beauloye · F. Zain · P. Malvaux · M. Maes (✉)
Unité d'endocrinologie pédiatrique, Département de Pédiatrie,
Cliniques Universitaires Saint-Luc, Avenue Hippocrate 10,
1200 Brussels, Belgium
e-mail: maes@pedi.ucl.ac.be
Tel.: +32-2-7641300; Fax: +32-2-7648910

J. Rahier · S. Gosseye
Department of Pathology,
Université catholique de Louvain, 1200 Brussels, Belgium

J. W. Honour
Department of Chemical Pathology and Endocrine Unit,
University College and Middlesex School of Medicine,
London, UK

V. Beauloye and F. Zain contributed equally to this work.

Lynn H. Gerber · Usha Chaudhry · Michael DeBaun

Joint laxity, scoliosis, and thoracic cage abnormalities in children with Beckwith-Wiedemann syndrome

Received: 13 June 2000 and in revised form 24 July and 28 August 2000 / Accepted: 28 August 2000

We report the frequency of joint laxity, scoliosis and thoracic cage asymmetry in 55 children with Beckwith-Wiedemann syndrome (BWS), findings not previously reported in the literature.

Beckwith-Wiedemann syndrome (BWS), originally reported by Beckwith [1, 2] and Wiedemann [5], described an overgrowth syndrome in which macroglossia, abdominal wall abnormalities (omphalocele), and gigantism were seen occurring together and serve as the basis for establishing the diagnosis. Other features reported include ear creases, facial naevi, hypoglycemia, renal abnormalities, and hemihypertrophy [5, 6]. Additionally, children with BWS have a high frequency of embryonal cancers in infancy and early childhood, which is associated with hemihypertrophy [4].

In an attempt to assess the possible associations between musculoskeletal abnormalities and cancer frequency in those with BWS, a systematic musculoskeletal examination was performed on 55 children. During 1993–1996 children with two or more of the following findings were enrolled in our study: (1) idiopathic, congenital macroglossia; (2) earlobe fissures; (3) umbilical anomaly; (4) birth weight and length greater than the 90th percentile; and (5) hypoglycemia (blood sugar <40 mg/dl) during the 1st week of life. The proband and all first-degree relatives underwent standardized assessment performed by two experienced physiatrists. The assessment described truncal features and recorded comparisons of right and left limb segment lengths and girths (Fig. 1). Upper and lower limb segment lengths were measured using standard bony landmarks, and girth was measured circumferentially, at mid-point of the limb segment using a tape measure. Asymmetry of length or girth was defined as $>10\%$ discrepancy between the right and left sides. Scoliosis was rated as present or absent by inspection of the thoracolumbar vertebral alignment and scapular prominence in standing and forward flexion. Thoracic cage asymmetry was present if nipple heights were discordant. The presence of pectus carinatum or excavatum were noted. Ligamentous laxity was considered present if the thumb reached the ventral forearm, or knees or elbows achieved more than 5° hyperextension, or calcaneus was everted more than 10° and medial arch was flattened.

There were 33 boys and 22 girls in the sample with a median age of 4 years (range 5 months to 13 years). A total of 38 children (70%) had laxity of one or more joints, 36 (65%) had thoracic cage asymmetry and 9 (16%) had scoliosis. There were 14 (25%) who had either pectus carinatum or excavatum abnormalities and 14 children (25%) had limb asymmetry with at least 10% difference in length or girth measurements.

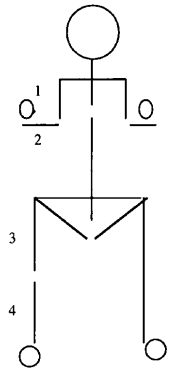

Fig. 1 Limb measurements. Limb lengths were measured from (1) acromioclavicular joint to olecranon, (2) olecranon to ulnar styloid, (3) anterior superior iliac spine to superior patellar border and (4) superior patellar border to medial malleolus. Limb girths were measured at the midproportion of the biceps brachii, midproportion of the brachialis, midproportion of the quadriceps and midproportion of the gastrocnemius

This study identified a high frequency of joint laxity and thoracic asymmetry, and to a lesser degree, scoliosis and chest wall deformities in a cohort of children with BWS. The original and subsequent reports of BWS have not reported these findings [2, 3]. Data on unaffected children are not available using the same careful systematic assessment, but it is the authors' clinical impression, based upon years of practice, that joint laxity is more prevalent in the BWS population.

The strong association between hemihypertrophy and cancer is well established [4, 6]. The observation that 25% of those with BWS have limb asymmetry, a 10% difference observed here, indicates that lateral differences do occur commonly in this population. The association of limb segment asymmetry (not hemihypertrophy) with the development of cancer needs to be tested in longitudinal observations. Joint laxity, thoracic cage asymmetries, to a lesser extent scoliosis, and limb length or girth asymmetry should be monitored over time to assess their functional impact and possible need for treatment.

References

1. Beckwith JB (1963) Extreme cytomegaly of the adrenal fetal cortex, omphalocele, hyperplasia of kidneys and pancreas, and Leydig cell hyperplasia – another syndrome? Annual Meeting of the Western Society for Pediatric Research. Los Angeles, Calif, Nov 11
2. Beckwith JB (1969) Macroglossia, omphalocele, adrenal cytomegaly, gigantism and hyperplastic visceromegaly. Birth defects V: 188–196
3. Elliot M, Bayly R, Cole T, Temple IK, Maher ER (1994) Clinical features and natural history of Beckwith-Wiedemann syndrome: presentation of 74 new cases. Clin Genet 46: 168–174
4. Miller RW, Fraumeni JF Jr, Manning MD (1964) Association of Wilms's tumor with aniridia, hemihypertrophy and other congenital malformations. N Engl J Med 270: 922–927
5. Wiedemann H-R (1964) Complexe malformatif familial avec hernie ombilicale et macroglossie, un syndrome nouveau. J Genet Hum 13: 223–232
6. Wiedemann H-R (1983) Tumours and hemihypertrophy associated with Wiedemann-Beckwith syndrome. Eur J Pediatr 141: 129

L. H. Gerber (✉) · U. Chaudhry
Department of Rehabilitation Medicine,
National Institutes of Health,
Building 10, Room 6S235,
10 Center Drive MSC 1604, Bethesda, MD 20892-1604, USA
e-mail: lynn_gerber@nih.gov
Tel.: +1-301-4964733; Fax: +1-301-4020663

M. DeBaun
Genetic Epidemiology Branch,
National Cancer Institute,
National Institutes of Health,
Bethesda, MD 20892-1604, USA, and
Washington University School of Medicine,
St. Louis, MO 63110, USA

Peter-Marc Fortune · Agostino Pierro · Andy Petros

Splanchnic oxygen delivery in exomphalos detected with near infrared spectroscopy

Received: 11 April 2000 and in revised form: 14 June 2000
Accepted: 25 August 2000

We report the use of near infrared spectroscopy to detect changes in oxyhaemoglobin signal from the abdominal viscera contained within a prolene silo in a neonate with exomphalos. Oxyhaemoglobin levels were low in the supine position and increased when the baby was turned onto either side.

Exomphalos is a rare congenital malformation of the umbilical cord alone or combined with a defect of the supra-umbilical region of the abdomen. Viscera herniate into a sac covered by amniotic membrane and peritoneum. In cases where a primary repair is not possible, the viscera can be enclosed within a prolene mesh silo which is then tightened to stretch the abdominal cavity to accommodate the viscera allowing formal closure [5]. There is an associated incidence of necrotising enterocolitis, believed to be due to splanchnic ischaemia secondary to raised intra-abdominal pressure [1]. Animal models have shown that the most profound reduction in perfusion occurs in the gut [3]. Splanchnic compromise can be estimated by measuring intra-vesical or inferior vena cava pressure [2].

Conventional management involves nursing the infant supine with the silo suspended vertically to encourage the gut to fall into the abdominal cavity. Currently there is no method to directly evaluate gut capillary bed oxygenation. Near infrared spectroscopy (NIRS) can determine changes in concentration of oxyhaemoglobin (HbO_2), deoxyhaemoglobin (Hb), total haemoglobin (tHb) and regional blood volume and has an established role in continuous, non-invasive, in-vivo monitoring cerebral oxygenation. Infra-red light was emitted and received from small optodes (Niro 500, Hamamatsu Photonics, Japan) placed on the right side of the silo and recorded a baseline signal for Hb, HbO_2 and tHb with the baby supine. NIRS has been applied through the abdominal wall and shown to detect changes in splanchnic oxygenation during periods of hypoxia in neonates [4].

We now report the use of NIRS to monitor splanchnic oxygenation in a 34-week gestation girl with an antenatal diagnosis of ruptured exomphalos. All abdominal viscera, except